EDITORIAL COMMENTARY



Screening for Congenital Hypothyroidism—Time to Raise the Bar?

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Screening for congenital hypothyroidism is essential to prevent the deleterious effect of the disorder. The European Society for Pediatric Endocrinology (ESPE) and American Academy of Pediatrics guidelines have been widely used for interpreting neonatal screening results [1, 2]. Consideration for regional factors and resource availability is mandatory to allow widespread implementation of the screening program. Indian Society for Pediatric & Adolescent Endocrinology (ISPAE) has recently provided guidance for screening, evaluation, and management of congenital hypothyroidism [3, 4]. These guidelines differ from the ESPE guidelines in the TSH action thresholds. The impact of this change on the diagnostic value of the screening strategy has not been studied.

Venugopalan et al. report, in this issue of the journal, their experience on the utility of ISPAE guidelines in identifying congenital hypothyroidism in a high-risk neonatal unit [5]. The retrospective chart review of 8064 neonates showed that the increase in screening TSH cutoffs as per the ISPAE guidelines decreased the detection rate of congenital hypothyroidism from 88.0% (73 out of 83) to 43.3% (36 of 83). This, however, did not impact the detection of congenital hypothyroidism, as all the 47 cases missed by the cutoff met the guidelines for rescreening and would have been identified on retesting. This is related to the "high risk" nature of the setting, as indicated by the need for retesting in 20.1% (1622 of 8064) neonates. The authors conclude that the upward revision of the screening TSH threshold is expected to lower the number of neonates recalled (increased specificity) while not missing cases (preserved sensitivity).

The study provides insights into the impact of revised cutoffs for neonatal screening. A disproportionately high representation of at-risk neonates and a prevalence of hypothyroidism (1 in 97) limit the generalizability of the study in the community. The retesting of 1 in 5 neonates in the study allowed detection of over half the neonates with congenital hypothyroidism missed due to revised TSH cutoffs. The diagnostic accuracy of revised TSH cutoff without the retesting safety net in the community remains unclear. This study confirms the applicability of ISPAE guidelines in an at-risk setting; the validation in the community awaits more extensive community studies.

Declarations

Conflict of Interest None.

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